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Genetic Testing for Breast Cancer Susceptibility: Awareness and Interest Among Women in the General Population

Ellen S. Tambor, Barbara K. Rimer,* and Tara S. Strigo
Duke Comprehensive Cancer Center, Durham, North Carolina

Previous research has suggested that demand for genetic testing for breast cancer susceptibility may be quite high, even among those at relatively low risk of carrying a mutation. This study examined the extent to which a group of female HMO enrollees were aware of the discovery of the *BRCA1* gene and, without having received detailed information about the test, whether they would be interested in being tested to find out if they have the gene. Factors associated with awareness of and interest in testing were also examined. Four hundred seventy-three women age 50 and over, almost all of whom did not have an increased risk of breast cancer based on family history, were surveyed by telephone. Fifty-one percent of respondents had heard about the discovery of a breast cancer gene. In logistic regression analysis, women who described themselves as comfortable financially, had at least some college education, and were premenopausal were more likely to have heard of the gene discovery than women who were not comfortable financially, had no more than a high school education, and were postmenopausal. Sixty-nine percent of the respondents said that they would be interested in being tested to find out if they had a breast cancer gene. Women who were younger than 60, white, believed their family would benefit if they had a mammogram, and believed that regular mammograms give them a feeling of control over their health, were more likely to be interested in testing than those who were 60 or older, African-American or other, and did *not* believe that their family would benefit if they had a mammogram or that mammograms give them a feeling of control over their health. These findings have implications for education and counseling. Women who express an interest in being tested must be made fully aware of the limitations and possible consequences of testing. Special efforts may be needed to make information about testing available to women who have low levels of education. Am. J. Med. Genet. 68:43–49, 1997 © 1997 Wiley-Liss, Inc.

KEY WORDS: genetic testing; breast cancer; BRCA1

INTRODUCTION

A genetic test for the *BRCA1* breast-ovarian cancer susceptibility gene [Futreal et al., 1994; Miki et al., 1994] is not yet widely available; however, several companies and centers have already begun to market testing [Beardsley, 1996], and widespread testing in the future is likely. *BRCA1* mutations are believed to occur in approximately 1 in 1,000–2,000 women in the U.S. and account for about 5% of breast cancer cases [Ford and Easton, 1995]. An 85% risk of breast cancer by age 70, along with a 63% risk of ovarian cancer, has been estimated for women with *BRCA1* mutations [Easton et al., 1995].

The prospect of more accurate and widespread identification of mutation carriers has focused attention on problems that may arise with testing and on the need to establish guidelines *before* testing is underway [American Society of Human Genetics (ASHG), 1994; Biesecker et al., 1993; King et al., 1993; Lerman et al., 1994a]. This process involves, in part, anticipating the demand for testing and attempting to characterize those women who are most likely to be interested in the test so that strategies for education and counseling can be developed.

Interest in genetic testing among family members of breast and ovarian cancer patients tends to be very high [Lerman et al., 1994b, 1995; Struewing et al., 1995], even among those who are not at a high risk of having a *BRCA1* mutation [Lerman et al., 1994b]. Women without a family history of breast and ovarian cancer have also shown a high level of interest in test-

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*Correspondence to: Barbara K. Rimer, Duke University Medical Center, Cancer Prevention, Detection, and Control, 2200 W. Main St. B-150, Durham, NC 27705.

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ing [Chaliki et al., 1995]. Conclusions about the demand for *BRCA1* testing based on such studies must, however, be interpreted cautiously due to the limited information that generally has been provided to participants concerning the risks and limitations of testing. It is likely that many women who show an initial interest in testing may decide against the test once they learn, for example, that following a negative test result they will still carry the general population risk for developing breast cancer.

Nevertheless, a substantial demand on medical and genetic resources may be produced by women who express an interest in testing even if they ultimately decide not to have the test. Achieving cost-effectiveness with genetic testing for breast cancer susceptibility is already anticipated to be very difficult [Brown and Kessler, 1995], especially for women who do not have a high probability of carrying a mutation. However, the massive education and counseling that will be required for those interested in testing could make it virtually impossible. This problem may be exacerbated by extensive media coverage surrounding the discovery of breast cancer genes and eventually the availability of genetic tests. It will be compounded by the limited number of genetic counselors.

The current study examines the extent to which a group of female HMO enrollees were aware of the discovery of the *BRCA1* gene and whether they would be interested in being tested to find out if they have the gene. As it was beyond the scope of this study, detailed information about the test was not provided prior to decision-making. Based on previous studies of interest in genetic testing for cancer susceptibility, we predicted that interest would be high [Chaliki et al., 1995; Croyle and Lerman, 1993; Lerman et al., 1994b, 1995; Smith and Croyle, 1995; Struewing et al., 1995]. In addition, we examined several potential predictors of awareness of breast cancer genes and interest in testing. We would predict, for example, that women with more education would be more likely to have heard about the gene discovery and will be more likely to become aware of test availability than women with less education. The variable most consistently associated with interest in genetic testing for cancer susceptibility in past research has been perceived risk, either of being a gene carrier or of developing cancer [Croyle and Lerman, 1993; Smith and Croyle, 1995; Lerman et al., 1994b; Struewing et al., 1995]. Interest in genetic testing for breast cancer might also be related to other health care attitudes and behaviors, particularly with regard to breast care [Chaliki et al., 1995].

MATERIALS AND METHODS

This study was conducted as part of a larger research project comparing the effectiveness of two interventions at increasing adherence to routine mammography. The project is a collaboration of Duke University Medical Center and Kaiser Permanente of North Carolina.

Sampling and Procedures

The survey sampling frame included women age 50 and older who were enrolled with Kaiser at one of five sites in the Raleigh-Durham-Chapel Hill area of North Carolina and who, according to Kaiser's records, had received two or fewer mammograms in the 3-year period from May 1991 to May 1994. In order to construct the cohort of women for intervention, we mailed eligible women a letter and consent card to return if they agreed to participate in the research project. In the first phase of the survey, women who, according to their own self-report, were adherers to mammography, as well as those who were self-reported nonadherers, were considered eligible to participate in the survey. In the second phase, only self-reported nonadherers were considered eligible. This variation in sampling was controlled for in subsequent analyses.

Eligible consenters were surveyed by telephone. All surveys were conducted by professional survey interviewers from Mathematica Policy Research with repeated attempts made to contact participants. Phase 1 of the survey was completed in October and November 1994 and Phase 2 was completed in February and March 1995.

Measures

The survey included the following two yes-no questions which were the primary outcomes for this study: (1) recently, scientists announced that they had found a gene that causes breast cancer. Did you hear about this?; and (2) if you could be tested to find out whether you had the gene, would you be interested? In addition, the survey included questions concerning: demographics, medical care and insurance, health behaviors, breast cancer risk, mammography history, mammography intentions, and attitudes and beliefs concerning mammography. Questions were either yes-no, multiple choice, or open-ended with precoded response categories. An "adherence to mammography" variable was constructed based on several survey questions concerning dates of previous mammograms. The survey took approximately 15–20 minutes to complete.

Analysis

In calculating response rates, women who became ineligible to participate in this research because they had died, moved with no forwarding address, were no longer Kaiser members, or had a double mastectomy were excluded from the denominator.

Chi-square statistics were computed to examine associations between the two outcome variables, awareness of gene discovery and interest in testing, and a number of sociodemographic, health behavior, risk factor, and attitudinal variables. Variables with significant chi-squares were entered into stepwise logistic regression models to predict the two outcomes.

Because changes were made to the sampling frame with respect to adherence to mammography, we took a couple of measures to control for the possible effect of this variation on the analysis. First, we looked for interactions between the adherence variable and all of the variables associated with awareness and interest. No interactions were found. Second, we forced adherence into all regression models before entering any other variables.

TABLE I. Sociodemographic Characteristics of Respondents

	% (N = 473)
Age (years)	
50–59	65
60–69	26
70+	9
Education	
High school or less	40
Some college	34
College graduate or more	26
Race	
White	83
Black	15
Other	2
Financial situation	
Very comfortable	87
Makes ends meet	9
Cut back/losing ground	4
Present working status	
Working	64
Not working	36
Marital status	
Married	66
Not married	34

RESULTS

Response Rates and Respondent Characteristics

Sixty-five percent of the women who were mailed consent cards agreed to participate in the research project. Of these, 82% of the eligible women completed the telephone survey (N = 473). The respondents' sociodemographic characteristics are presented in Table I. The sample is predominantly white and the majority of respondents were between 50 and 59 years of age. Only 26% of the sample graduated from college, but most described themselves as very comfortable financially. The majority of the women were married and currently working.

Only 6% of the respondents reported that their mother had been diagnosed with breast cancer and of these only 14% had been diagnosed before age 50. An additional 4% had a sister with breast cancer, 39% of whom had been diagnosed before age 50. Only 2% of the sample could be considered at increased risk for breast cancer on the basis of family history. There was a fairly even split between women who were self-reported adherers to routine mammography (44%) and those who were nonadherers (56%).

Awareness of Breast Cancer Gene

Fifty-one percent of respondents reported that they had heard about the discovery of a gene that causes breast cancer. Table II shows the variables that were associated with awareness in bivariate analysis. The results of the logistic regression analysis are presented in Table III. Women who said that their financial situation is comfortable were over four times as likely to have heard of the breast cancer gene as women who said they were not doing well financially. Respondents

TABLE II. Factors Associated With Awareness of Breast Cancer (BC) Gene*

	Heard of BC gene % (N = 236)	Have not heard of BC gene % (N = 224)
Education		
High school or less	28	52
Some college	38	31
College grad or more	34	18
Financial situation		
Comfortable/enough	99	93
Cut back/losing ground	1	7
How many babies have you had?		
None	7	11
1–4	82	69
>4	11	20
Stopped getting menstrual period		
Yes	81	90
No	19	10
Ever had breast problem		
Yes	39	28
No	61	72
Number of mammograms		
None	6	8
1–2	18	28
3 or more	77	64
Frequency of clinical breast exam		
At least every 3 years	87	79
Less than every 3 years/no sched.	13	21
Mammograms lead to unnecessary breast surgery		
Agree	5	11
Don't agree	95	89
Having a mammogram is looking for trouble		
Agree	3	7
Don't agree	97	93

* All Ps $< .05$.

TABLE III. Logistic Regression: Awareness of Breast Cancer Gene

Variable	Parameter estimate	Standard error	Odds ratio	*P* value
Adherence to mammography	.3542	.1984	1.425	.074
Financial situation (comfortable)	1.4899	.6564	4.437	.023
Education (high school or less)	−.9566	.2031	.384	<.001
Stopped menstrual period	−.5624	.2855	.570	.049

with no greater than a high school education were about 2.5 times less likely to have heard about the gene than those with at least some college. Finally, those who had not stopped getting their menstrual periods were more likely to be aware of the gene discovery than postmenopausal women.

Interest in Genetic Testing for Breast Cancer

Sixty-nine percent of the women surveyed said that they would be interested in being tested to find out if they have a breast cancer gene. Twenty percent said that they would not be interested in testing and an additional 10% said they did not know. Variables that were significantly associated with interest in testing are presented in Table IV. In the logistic regression model (Table V), women younger than 60 were almost three times as likely to be interested in the test as women age 60 and over, and white women were over twice as likely to be interested as African American and other women. No other sociodemographic variables were significant predictors of interest in testing in the model. In addition, women who believed their family would benefit if they have a mammogram were twice as likely to be interested as those who did not believe their family would benefit and women who believed that having regular mammograms gives them a feeling of control over their health were almost three times as likely to be interested in genetic testing as those who did not agree that mammograms give them a feeling of control.

DISCUSSION

Factors Associated With Awareness and Interest in Testing

About half of the women we surveyed had heard about the discovery of a gene that causes breast cancer. As hypothesized, women with higher educational attainment and a comfortable financial situation were more likely to be aware of the gene discovery than those with less education and a less comfortable financial situation. Women in these groups may be more likely to pay attention to and have access to sources of information regarding advances in genetic technology. We also found that, even though there was no association between gene awareness and age, premenopausal women were more likely to have heard of the gene discovery than women who had stopped getting their menstrual periods. Perhaps, postmenopausal women are more concerned about other health issues. Alternatively, they may be very concerned about breast cancer in general but less concerned about genetic factors.

Although the majority of the women we surveyed said that they would be interested in genetic testing for breast cancer (69%), the level of interest was substantially lower than has been found in other studies of interest in genetic testing for cancer susceptibility [Chaliki et al., 1995; Croyle and Lerman, 1993; Lerman et al., 1994b, 1995; Smith and Croyle, 1995; Struewing et al., 1995]. That this population of women was less interested in testing than relatives of breast and ovarian cancer patients is not surprising; however, levels of interest as high as 83% and 90% have also been reported in studies of the general population [Chaliki et al., 1995; Croyle and Lerman, 1993; Smith and Croyle, 1995]. Given that interest in testing was inversely related to age in this study, the discrepancy may be attributable to the fact that our study population consisted only of women age 50 and older, whereas other studies represented a broader age range. Our finding that older women were less likely than younger women to be interested in genetic testing is consistent with previous research [Lerman et al., 1994b]. Older women may be more concerned about their age as a cancer risk factor than about a possible genetic predisposition. In addition, they are more likely to be already engaged in the breast cancer screening practices that would be recommended for a woman found to have a mutation.

The finding that white women were significantly more likely to be interested in testing than African-American women is not supported by previous studies of interest in genetic testing for cancer susceptibility [Chaliki et al., 1995; Croyle and Lerman, 1993; Lerman et al., 1994b, 1995; Smith and Croyle, 1995; Struewing et al., 1995]. However, past studies have included very few African Americans or did not report data concerning race. Several explanations for this association are possible. For example, African-Americans tend to perceive their risk of cancer as lower than do whites [EVAXX, 1981], and may therefore see little benefit in genetic testing for breast cancer. Furthermore, African-American women have limited awareness of the increased risk associated with a family history of breast cancer [Audrain et al., 1995; Royak-Schaler et al., 1995]. Alternatively, black women may have a more negative view of genetic testing in general due, perhaps, to earlier experiences in this country surrounding genetic testing for sickle cell anemia [Markel, 1992]. In any case, these results suggest that there may be a need to communicate ethnically sensitive information about breast cancer risk and the availability of genetic testing for African-American women at potentially high risk for breast and other cancers.

We hypothesized that interest in genetic testing for breast cancer might be associated with attitudes and behaviors concerning mammography, as has been

TABLE IV. Factors Associated With Interest in Genetic Testing for Breast Cancer*

	Interested in testing % (N = 319)	Not interested in testing % (N = 141)
Age (years)		
50–59	72	48
60–81	28	52
Race		
White	87	80
Black	13	20
Working status		
Working	68	55
Not working	32	45
Adhere to mammography schedule		
Yes	47	35
No	53	65
Had clinical breast examination in past two years		
Yes	86	77
No	14	23
Intentions regarding mammogram		
Planning/thinking about having	97	86
Not planning	3	14
How often plan mammogram in future		
On schedule (1–2 yrs)	86	74
Not on schedule	14	26
Family benefits if have mammogram		
Agree	91	74
Don't agree	9	26
Mammograms cause worry about breast cancer		
Agree	13	23
Don't agree	87	77
Mammograms give feeling of control over health		
Agree	89	71
Don't agree	11	29
After normal mammograms, can skip a few years		
Agree	7	15
Don't agree	93	85
No mammogram unless have breast problem or pain		
Agree	6	11
Don't agree	94	89
Mammograms give peace of mind about health		
Agree	91	81
Don't agree	9	19
Need mammogram even if no family history of breast cancer		
Agree	97	87
Don't agree	3	13
Would put off mammogram because of pain involved		
Agree	7	13
Don't agree	93	87

* All Ps $< .05$.

demonstrated previously [Chaliki et al., 1995]. A number of variables related to health care behaviors and intentions, such as recency of clinical breast exam and plans for future mammograms, were associated with interest in testing in bivariate analysis but were not significant predictors in the regression analysis. Attitudes toward mammography, however, were important predictors of interest in testing. Specifically, women who said that their family would benefit if they had a mammogram were more likely to be interested in genetic testing than those who did not believe their family would benefit. Although concern for family in this context implies a belief that by taking care of their own health they are benefiting their family, it might also indicate a desire to acquire information which might be useful to other family members, such as a daughter. Data from focus groups we recently conducted suggest that helping family members is a strong motivation for genetic testing. Women who reported that having regular mammograms gives them a feeling of control over their health were more likely to be interested in testing than those who did not believe that mammograms give them a feeling of control. The potential knowledge acquired through genetic testing and the subsequent improvement in decision-making ability might appeal to individuals who are motivated by a desire to maintain control over their health.

Like others [Lerman et al., 1994b, 1995; Struewing et al., 1995], we did not find an association between actual risk factors, such as having a mother who had been diagnosed with breast cancer, and interest in genetic testing for breast cancer. The general conclusion has

TABLE V. Logistic Regression: Interest in Genetic Testing

Variable	Parameter estimate	Standard error	Odds ratio	*P* value
Adherence to mammography	.1994	.2339	1.221	.39
Age (<60 years)	1.0801	.2274	2.945	<.001
Race (white)	.8071	.2986	2.241	.007
Family will benefit if have mammogram	.7260	.3375	2.067	.03
Regular mammogram gives feeling of control over health	1.0268	.3196	2.792	.001

been that perceived risk, which often does not coincide with actual risk, is the more important determinant of interest in testing. However, we also failed to find a significant association between perceived risk and interest in testing. This result may be due to the wording of the question we used to measure perceived risk (developed for other research purposes) which asked participants to indicate whether they believed they were less likely than other women their age to get breast cancer. The real difference may be between women who believe they are at low to average risk and those who believe they are more likely than other women their age to get breast cancer.

Implications for Education and Counseling

As suggested by these findings, the education and counseling needs of women who are interested in genetic testing for *BRCA1* are likely to vary. For example, young women may have different concerns than older women, and women with children may have concerns that do not apply to those without children. African-American women may have their own unique set of concerns. As new discoveries about *BRCA1* and other breast cancer genes are made, education and counseling will become even more complex. For example, the discovery of a strong association between the 185delAG mutation and early onset breast cancer in Ashkenazi Jewish women [Fitzgerald et al., 1996] introduces additional information that is relevant to a specific demographic group.

Given the complex education and counseling requirements that accompany genetic testing for cancer susceptibility, strategies to augment traditional genetic counseling approaches should be pursued. We are currently testing a strategy of providing printed educational materials prior to a genetic counseling session that are tailored for each participant based on her responses to a baseline survey. So, for example, only women who have or are planning to have children will receive detailed information about the implications of testing for one's children, and only Jewish women will receive information on the 185delAG mutation. Tailored messages will also highlight individual knowledge deficits and concerns about testing. We hope that by providing personalized informational materials, women will become more engaged in the decision-making process at an early stage, thereby enhancing the overall education and counseling process. The use of tailored messages in printed educational materials represents one example of how education and counseling for genetic testing for cancer susceptibility might be improved.

Limitations

A number of limitations to this study should be noted. First, the survey participants represent a fairly select population of older, predominantly white women, the majority of whom described themselves as "very comfortable" financially. However, it should be noted that the HMO includes a wide range of incomes. This question reflects respondents' perceptions of how they are coping financially. Secondly, because this study was conducted as part of a larger research project, we were unable to provide much information about genetic testing for breast cancer. It would be particularly interesting to assess possible changes in decision-making following disclosure of information about the test. Finally, we were not able to adequately answer certain research questions (i.e., the effect of perceived risk on interest in testing), because the survey items were developed for other research purposes.

Conclusions

Given the high level of initial interest in genetic testing for breast cancer found in this study, and the even higher levels reported by other studies, it is important to ensure that women who express an interest in being tested are made fully aware of the limitations and possible consequences of testing. Furthermore, because women at higher socioeconomic levels tend to be more aware of genetic discoveries than those at lower socioeconomic levels, an effort should be made to provide information about and access to testing to all women who might benefit from and be interested in testing once it becomes available. Our findings suggest that interest in testing is related to demographic characteristics as well as to beliefs and attitudes concerning health behaviors and breast cancer. Further research is needed to gain a better understanding of these associations. In addition, research to test different approaches to educating and counseling women about genetic testing for cancer susceptibility is greatly needed.

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